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ORIGINAL ARTICLE

Users' motivations to purchase direct-to-consumer genome-wide testing: an exploratory study of personal stories

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Abstract The relatively rapid growth of the direct-to-consumer (DTC) genetic testing market in the last few years has led to increasing attention from both the scientific community and policy makers. One voice often missing in these debates, however, is that of the actual user of these genetic testing services. In order to gain a better picture of the motivations and expectations that propel individuals to purchase DTC genome-wide testing, we conducted an exploratory study based on users' personal stories. Through qualitative content analysis of users' personal stories found on Internet blogs and DTC genetic testing companies' websites, we identified five major sets of motivations and expectations towards DTC genome-wide testing. These themes are related to (1) health, (2) curiosity and fascination, (3) genealogy, (4) contributing to research, and (5) recreation. Obtaining such information can help us to understand how users consider genome-wide testing and forms the basis for further research.

Introduction

New genomic knowledge is shifting the focus from testing for specific and rare monogenic diseases to genetic and genomic testing to predict the risk of developing common complex diseases. The current advances provide the hope to develop new methods in prediction, prevention, diagnosis, and treatment of diseases and of achieving better health in general (Khoury 2003). The translation of genomic medicine is an essential goal for the medical and public health community; however, the effective transfer of emerging genomic knowledge and applications into clinical settings is a long process which has to overcome many barriers.

Elaborating on the results of genome-wide association studies, some private companies have been advertising and selling genetic tests directly to consumers (Hunter et al. 2008). As a consequence, consumers can now obtain genetic tests without any involvement from a health care professional. The companies selling these services argue that obtaining genetic information can help individuals to construct their identity (by ancestry testing) or promote their health (by changing their habits after having received disease risk predictions). According to these companies, after receiving personal risk predictions based on genetic information, consumers will be able to use the test results in their daily life, particularly in monitoring or improving their health conditions (Foster et al. 2006). Autonomy, empowerment, prevention, convenience, and privacy are the principle keywords in the marketing of these direct-to-consumer (DTC) genetic tests (Howard and Borry 2009).

The rapid growth of the DTC genetic testing market has led to increasing attention from both the scientific community and policy makers. Different types of genetic tests sold directly to consumers (i.e., carrier testing versus testing for common complex diseases or single-gene testing versus testing for hundreds of different loci) raise different specific scientific, ethical, legal, and social concerns; however, a general list of some of the important concerns regarding the field in general can be outlined. Concerns have been raised

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about the scientific accuracy, clinical validity, and clinical utility of DTC genetic tests, especially for common complex traits (Janssens et al. 2008; Mihaescu et al. 2009; Foster et al. 2009). Concerns were also raised with regard to the absence and/or quality of the pre- and posttest genetic counseling (Wade and Wilfond 2006), as well as with the absence of individualized medical supervision (Hogarth et al. 2008). The lack of an adequate consent procedure as well as the inappropriate genetic testing of minors has also been criticized (Borry et al. 2009a, b). Further concerns include the research activities of DTC genetic testing companies (Howard et al. 2010), the (lack of) respect for privacy, and the potential burden on public health resources (McGuire et al. 2009).

Various professional organizations and governmental agencies have published their statements to inform, educate, and/or warn consumers about DTC genetic testing (American College of Obstetricians and Gynaecologists 2008; Federal Trade Commission 2009; Human Genetics Commission 2010; United States Government Accountability Office 2006; United States Government Accountability Office 2010; Nuffield Council on Bioethics 2010). Along these lines, the European Society of Human Genetics endorsed in August 2010 a statement which included, among others, recommendations to ensure the quality of the testing services, the provision of pretest information and genetic counseling, a face to face consultation, and oversight of this industry (European Society of Human Genetics 2010). Moreover, since the spring of 2010, the US Food and Drug Administration has decided to investigate more closely the market activities of DTC genetic testing companies, which may impact on the future regulatory oversight of the DTC genetic testing market (Allison 2010).

At the present time, little information is known about the motivation of users to purchase DTC genetic tests. McGowan and colleagues (McGowan et al. 2010) published the results of a qualitative study in which they interviewed 23 early users (those who purchased tests "within the first two years of their availability on the market") of personal genome services. To date, this is the only full-length publication providing firsthand accounts of DTC genetic testing (GT) users' reasons for purchasing DTC personal genome services. When asked about the main reasons for purchasing personal genome services, the interviewees most frequently answered that they did so to: (1) "gain health-related information" and (2) "learn about individual risk factors." Additional motivation factors included wanting to be at the "vanguard of adopting and testing the capabilities of new technologies" and having a professional interest in the service (McGowan et al. 2010). Kaufman and colleagues also presented the results of an online survey of 1,048 customers of three direct-to-consumer personal genomic testing companies at the American Society of Human Genetics 60th Annual Meeting (2010). (Kaufman et al. 2010). Although the full article has yet to be published, the abstract reveals that the top three reasons for customers to purchase DTC genetic testing were identified as (1) satisfying curiosity, (2) learning about elevated risks of diseases, and (3) learning about ancestry (Kaufman et al. 2010). Other studies have looked at the attitudes of members of the general public regarding genetic testing and the provision of these tests directly to consumers (Wilde et al. 2010). One study (McGuire et al. 2009) surveyed the general public via facebook to find out, among others, about their use and interest in genetic testing, as well as their attitudes towards DTC genetic testing companies. Yet, other studies created circumstances that were similar to the DTC genetic testing context, but that were still different in a number of ways. Bloss et al. (2010) recruited mainly employees from a number of health and technology companies and invited them to take part in a study whereby participants were offered genome-wide testing from a DTC genetic testing company at a significantly discounted rate compared with what individuals in the general public would have to pay. They reported primarily on demographic characteristics of the participants as well as concerns participants had regarding being involved in the project and obtaining results for an unpreventable disease (Bloss et al. 2010). Meanwhile, through the Multiplex initiative, McBride et al. (2009) tried to evaluate what psychological and behavioral factors predict who is likely to seek SNP-based genetic tests for multiple common health conditions. In addition, a form of personal case study has also been published by at least two academics and one journalist who purchased DTC genetic testing. Richards (2010) published an account of his experience in a peer-reviewed journal, and Collins and Duncan each discussed their experiences as a part of full-length books (Collins 2010; Duncan 2009).

Other than the studies conducted by McGowan et al. (2010) and Kaufman et al. (2010), the abovementioned studies provide information about a type of public concerning genetics and/or genomic testing but were not focused on an actual population of users of DTC GT. Herein, we specifically wanted to obtain information from individuals who have already purchased genome-wide testing sold by a DTC genetic testing company. Our goal was to explore their motivations for purchasing these tests and to further understand their expectations of these services. To do so, we collected and analyzed (via Internet postings) users' personal stories regarding their experiences with purchasing DTC genome-wide testing.

Methods

Definitions

Genome-wide testing: For the purposes of this study, the term genome-wide testing (GWT) is used to mean the testing of hundreds of thousands to millions of genetic markers (often single nucleotide variants (SNPs)) throughout the genome. Terms such as "full genome testing" (Howard et al. 2010), "personal genome scanning" (McGowan et al. 2010), or "personal genomic testing" (Cherkas et al. 2010) have also been used to denote this type of testing. Companies selling DTC GWT typically return personalized risk estimates/profiles for multiple health conditions, and/or for pharmacogenomic information, and non-health related traits (i.e., eye color, hair type); some companies also return genealogical information. Companies that offer GWT are a subset of DTC genetic testing companies. That is to say that not all DTC GT companies sell GWT; some companies only analyze one or a few genetic variations.

User or customer: The focus population of this study are individuals who have already purchased and used a genome-wide test from a DTC GT company. In order to distinguish an actual user of a genetic test from a general consumer who may or may not purchase a genetic test, we use the terms "user" and "customer" to denote the former.

Data collection

The personal stories of customers who purchased DTC GWT for themselves and/or for their families were collected for this study. The aim was to find stories of actual users explaining their motivations and expectations for having purchased DTC GWT. Such stories were found on non-company websites as well as on DTC GWT companies' websites (which post customer stories). Non-blog mass media news items, in which reporters discussed their (or other customers') experiences related to DTC genetic testing, were excluded from this study. Moreover, due to the complex methodological issues raised by the analysis of images (Silverman 2006), video segments (e.g., Youtube) were also excluded.

Customer stories were collected via the Internet search engine Google. Five companies which provide DTC genome-wide testing services were selected as targeted companies in this research: 23andMe, deCODE (service called deCODEme), Navigenics, Seqwright, and Biomarker Pharmaceutical (service called Gene Essence). As a search strategy, the keywords "My DNA result" + "company name" or "service name" were used in Google search during the time period from February 17 to 20, 2010. As this study was aimed to be an explorative study, not all stories found were analyzed; a convenient sample of 120 stories was selected for preliminary analysis. Of the stories obtained in this way, only stories written in English and in which users addressed their reasons to purchase the test and/or expectations of the tests were kept for content analysis.

Data analysis

Stories were analyzed using content analysis. Words, sentences, and paragraphs that described customers' expectations or motivations were collected in the preliminary analysis to develop codes. At the early stage of analysis, the original words from these stories were used as code labels, and a preliminary coding taxonomy was generated during the investigation. A second review of the stories and the coding taxonomy helped to create advanced categories to conceptualize customers' detailed expectations and motivations towards DTC genetic testing. Themes and subthemes were developed under each advanced category.

Results

Customers' stories

Of the 120 stories obtained for preliminary analysis, 56 stories were written in English and addressed DTC GWT users' expectations and motivations for purchasing tests. These 56 stories were, therefore, retained for content analysis. Twenty-nine (52%) stories were published on DTC GWT companies' websites (often in the form of an interview), and 27 (48%) stories were found on non-company websites (usually in the form of a first-person blog).

Based on the personal information provided by customers in their stories, it would appear that the 56 stories studied were posted by 47 different individuals; five customers blogged more than once about their experience (on non-company websites), and each blog story was counted as a different story. Furthermore, due to the fact that some customers purchased more than one test from more than one company, the 56 stories represent the purchasing of 60 genome-wide tests from three different companies. No customer stories describing motivations and expectations were found in which Gene Essence or SeqWright are mentioned. The majority of users in our study purchased genome-wide tests from 23andMe (57%; 34/60), 30% (18/60) of tests were purchased from deCODE (deCODEme), and 13% (8/60) from Navigenics. Both 23andMe and deCODE offer, among others, health-related results and ancestry or genealogy information, while Navigenics only offers health-related results. The 56 stories studied were published between December 2007 and January 2010; all stories were

first accessed in February 2010 and verified again in September 2010.

Some educational and/or professional information could be retrieved for 38 of the 47 customers: 8 are trained in genetics or medicine, 15 are businessmen, entrepreneurs, attorneys, or engineers, and 15 are journalists or professional bloggers. Nineteen out of 47 customers identified themselves as parents, and considered taking the test not only for themselves but also for their families.

Themes and subthemes

Based on our analysis, users of DTC GWT expressed five predominant sets of motivations and expectations towards DTC genome-wide testing (Table 1). These themes are related to (1) health, (2) curiosity and fascination, (3) genealogy, (4) contributing to research, and (5) recreation. Some quotes from customers are relevant for more than one (sub)theme. Due to the possibility that themes obtained from stories on company websites could differ from themes found in stories obtained on non-company websites, we evaluated both groups separately as well as together. We found that stories from both origins express the same themes. However, in stories found on company websites, the most cited reason by far to purchase DTC GWT is health-related. Meanwhile, in stories from non-company websites, the most important reasons to purchase tests were curiosity and genealogy.

Theme 1: health-related motivations and expectations

Overall, the foremost reason to purchase DTC GWT is associated with health-related motivations and expectations. However, as stated above, this theme was much more predominant in stories from company websites, while it was much less important in stories found on non-company websites.

Subtheme 1.1: to address concerns for specific diseases Some customers expressed their concerns for a specific disease as a motivation to purchase DTC GWT. They often referred to the fact that "this" disease runs in the family. Various customers expressed that they want to know their own risk for developing that specific disease.

> I chose to take the deCODEme complete scan because I also wanted to learn about my genetic predisposition for developing age-related macular degeneration (AMD). My grandmother developed AMD in her mid 70s and, for years, I have wondered if I would one day be among the 40% of Canadians over the age of 75 to develop AMD. (source: company website, Anna Peterson, deCODEme: http://www.decodeme.com/customer-stories/the-gift-of-knowledge-genetics-and-prevention, accessed 24 September 2010)
>
> The genetic osteoporosis problem was a major reason why I decided to get a DNA SNP scan. (source: non-company website, Jerry Emanuelson, deCODEme and 23andMe: http://www.futurescience.com/dna/je-comments.html, accessed 24 September 2010)
>
> "One of the reasons that I was so interested in having genetic testing is my family history. Having someone in your life with Alzheimer's is a really tough thing. Being able to find information about my genetic risk factors that would allow me to take an active role in managing my health was very appealing to me." (source: company website, Jim, Navigenics: http://www.navigenics.com/visitor/what_we_offer/success_stories/jim/, accessed 24 September 2010)

Subtheme 1.2: to improve the quality of health and to live longer Some users expressed the belief that their genetic risk information would empower them to make positive future healthcare decisions, improve their health, and allow them to live a longer life.

Table 1 Overview of themes and subthemes obtained from customers' stories regarding direct-to-consumer genome-wide testing

Theme 1: health-related motivations and expectations[a]
Subtheme 1.1: to address concerns for certain diseases
Subtheme 1.2: to improve the quality of health and to live longer
Subtheme 1.3: to learn about medical history
Theme 2: motivations and expectations related to curiosity and fascination[b]
Subtheme 2.1: curiosity and fascination about the technology
Subtheme 2.2: curiosity and fascination about genetic information generated and learning about genetics/genomics
Theme 3: genealogy-related motivations and expectations[b]
Theme 4: motivations and expectations related to participation in or contributing to research
Theme 5: recreation-related (non-genealogy) motivations and expectations

[a] the predominant theme obtained from customer stories found on company websites

[b] the predominant themes obtained from customer stories found on non-company websites

> Personally I think having more information about myself can only be a good thing. Because you can act on it. A gene coding for a prostate cancer predisposition isn't a death sentence—it's a call to action. Eat better, get exercise, get checked every year after you're 40. That sort of thing. (source: non-company website, Nat Friedman, 23andMe: http://nat.org/blog/2008/02/personal-genetics/, accessed 24 September 2010)
> "By learning about my genetic predisposition for different illnesses, I will be better prepared to take an active role in my future health care decisions." (source: company website, Anna Peterson, deCODEme: http://www.decodeme.com/customer-stories/the-gift-of-knowledge-genetics-and-prevention, accessed 24 September 2010)
> "I almost feel evangelical about my health," says the 62-year-old businessman from Spokane, Washington. "I'm so excited about what can happen if you do the right things and have the right tools." (source: company website, Jack Doughery, deCODEme: http://www.decodeme.com/customer-stories/genetic-test-is-sensible-self-investment, accessed 24 September 2010)
> "I wanted to participate in Navigenics' service, because I was interested to see what I could learn about my health. I wanted to know what conditions I might be predisposed for in the future, so that I could take steps to protect myself." (source: company website, Jackie, Navigenics: http://www.navigenics.com/visitor/what_we_offer/success_stories/jackie/, accessed 24 September 2010)

In parallel with the previous subtheme, some customer stories from company websites expressed the motivation to learn about potential health risks for their children.

> And at a more practical level, I want to give my kids as much information as possible about the cocktail they are inheriting from my husband and me, so they can better manage their health. (source: company website, Kim Scott, 23andMe: https://www.23andme.com/pregnancy/profile/KimScott/, accessed 24 September 2010)
> Maybe learning about my own genetic make-up will give me the information I need to keep my children healthy, help them grow. (source: company website, Nancy Rabinowitz-Friedman, 23andMe: https://www.23andme.com/pregnancy/thread/708/, accessed 24 September 2010)
> "I wanted to know my risks for my daughter's sake. She shares my genes. She comes from me. So I'm interested to see what her risks are. If they are increased because of me. It could save her life," Pam explains. (source: company website, Pam Bale, deCODEme: http://www.decodeme.com/customer-stories/dna-test-perfect-christmas-present, accessed 24 September 2010)

Subtheme 1.3: to learn about medical history Some customers expressed the wish to obtain information about their medical history through DTC GWT. They considered this a way to substitute for their missing medical history and obtain medical information that was not provided to them by family members (e.g., because of adoption).

> For me, it's deeply personal. I don't have a medical history. I can't tell you who my ancestors were. I was adopted…I've never really understood what doctors mean by that final "I see," but I think it means, "Good luck with the crap shoot that is your medical history." So I've taken the test. I've sent my saliva to California to determine what's inside me. (source: non-company website, Jay Mueller, 23andMe: http://www.3aw.com.au/blogs/3aw-breakfast-blog/jays-dna-blog/20081209-6up0.html, accessed 24 September 2010)
> Also, because my father is a pathological and absent liar, I thought it would be cool to learn a bit about what diseases or conditions I might be pre-disposed to. Certainly, I can't depend on anything he has told me. (source: non-company website, Wendie Tobin, 23andMe: http://mommytopia.com/2009/01/13/23andme-sucks-ok/, accessed 24 September 2010)
> In the health and medical world, the knowledge base has grown so much since my parents died. My dad has been gone since 1992 and my mom died in 2000. My siblings and I don't have our parents, so being able to spit into a cup and find some of the answers to our family health history is a blessing and a tremendous opportunity. (source: company website, Michelle Lamar, 23andMe: https://www.23andme.com/pregnancy/thread/714/, accessed 15 January 2011)

Moreover, on company websites, some users expressed the value of sharing this family history information with their relatives in order to provide them with information about their health.

> "I was adopted, so I don't have any specific information about my family history. None of that information is available to me. That has always concerned me."(…) "Now I can give my daughter information about her own health. It's so thrilling – for both of us – to go through the Navigenics testing and see my results. Suddenly we have so much more

information than most people have about their genetic make-up." (source: company website, Terry, Navigencis: http://www.navigenics.com/visitor/what_we_offer/success_stories/terry/, accessed 24 September 2010)

The more we know about our family history, the more we know about ourselves. I spat and became part of the 23 and Me community because I want to know more about my ancestors, so I can help myself and my family live longer, healthier lives. (source: company website, Michelle Lamar, 23andMe: https://www.23andme.com/pregnancy/thread/714/, accessed 24 September 2010)

Theme 2: motivations and expectations related to curiosity and fascination

Various users expressed a fascination for genetics/genomics science and for the technology or information offered through DTC GWT (that is not based specifically on obtaining health or genealogy information). This was the predominant theme encountered in blogs found on non-company websites.

Subtheme 2.1: curiosity and fascination about the technology

Like many early adopters, my interest in sequencing my genome is driven more by intellectual curiosity than by pressing health reasons. So I'm hoping that George Church gets his Personal Genome Project geared up quickly. I've registered to participate. (source: non-company website, Gary Wolf, 23andMe: http://www.kk.org/quantifiedself/2008/01/-like-other-early-23andme.php, accessed 24 September 2010)

It will be interesting to see how useful this is. I'm fascinated with genetics, and when whole genome sequencing becomes affordable, I'll definitely do that. The technology is moving quickly. (source: non-company website, Mark Fletcher, 23andMe: http://wingedpig.com/2007/12/06/23andme-unboxing/, accessed 24 September 2010)

We are curious consumers and enjoy the chance to learn a little more about ourselves no matter how young and immature the personal genomics space is. (source: non-company website, Andrew Meyer, 23andMe: http://buzzyeah.com/2008/04/24/how-heart-attack-and-other-four-star-rated-topics-relate-to-my-dna-part-2/, accessed 24 September 2010)

"I just think it's interesting to see what the results are. To me it's fantastic deCODE can do it." (source: company website, Cheryl Click, deCODEme: http://www.decodeme.com/customer-stories/the-genetic-lottery, accessed 24 September 2010)

For some, this fascination is related to their professional backgrounds and interests.

My work involves building new computer programs that can help doctors and patients collect, track, understand, and use family health history and genetic/genomic information in patient care. Therefore I felt it made perfect sense that I should explore how technology – both gene chips (or DNA microarrays) and the Internet – could combine to give me access as never before to my own biology. (source: non-company website, Grant Wood, 23andMe: http://exploringmygenes.blogspot.com/2009/04/influences-on-my-decision.html, accessed 24 September 2010)

So why did I finally decide to go through with it? One reason is that I get a lot of questions from people at the Ask a Geneticist site about how useful or good the test is. Right now I have to tell them I don't know. I'd like to be more helpful than that. (source: non-company website, Barry Starr, 23andMe: http://www.kqed.org/quest/blog/2009/07/20/taking-the-plunge-diving-into-my-dna/, accessed 24 September 2010; according to the blog, Dr. Barry Starr is a Geneticist-in-Residence at The Tech Museum of Innovation in San Jose, CA and runs their Stanford at The Tech program.)

Some customers purchased tests because they expect this is the way medicine will evolve in the future.

I am a bit on the fence about this since it may just give me more to worry about but I also believe that this is the future of medicine. (source: non-company website, Avram Miller, Navigenics: http://twothirdsdone.com/2007/12/, accessed 24 September 2010)

"We hear a lot of different – and sometimes conflicting – opinions about how to take care of your heath. I'm very excited about a future in which I can receive only the most relevant information to me, based on my DNA. With these tests there will be an amazing amount of much smarter and more effective treatments. There are so many things that medicine can do better if they know enough about you to really target what they are doing." (source: company website: Aaron, Navigenics: http://www.navigenics.com/visitor/what_we_offer/success_stories/aaron/, accessed 24 September 2010)

"I believe it is the way of the future. Even ten years ago, who would have thought that we would have all the advances we have today? People are often scared of things just because they're new. It's about how we educate people and how we make them feel about it. They need to feel comfortable with it." (source: company website, Pam Ayers, deCODEme:

http://www.decodeme.com/customer-stories/dna-test-perfect-christmas-present, accessed 15 January 2011)

Subtheme 2.2: curiosity and fascination focused on the genetic information generated and learning about genetics/genomics Various users expressed a fascination about the genetic knowledge generated by DTC genetic testing.

I'm fascinated by genetics and am excited to learn more about what makes me tick. (source: company website, Cecily Kellogg, 23andMe: https://www.23andme.com/pregnancy/profile/Cecilyk/, accessed 24 September 2010)

"Being an engineer, when I found out I could take these tests through Navigenics and really understand more about my genetics – I was fascinated and really wanted to do it." (source: company website, Aaron, Navigenics: http://www.navigenics.com/visitor/what_we_offer/success_stories/aaron/, accessed 24 September 2010)

Being a daily witness to my family's health issues has made me incredibly curious about genetics and what traits and medical ailments could possibly be passed on to my son. (source: company website, Calliopeblogger, 23andMe: https://www.23andme.com/pregnancy/profile/Calliopeblogger/, accessed 24 September 2010)

Because of our imminent mortality, it is natural for us to ask whether we will learn something about our futures from these types of tests. (source: non-company website, Grant Wood, 23andMe: http://exploringmygenes.blogspot.com/2009/04/influences-on-my-decision.html, accessed 24 September 2010)

Some customers expected that the genetic information could provide insights into their identities and what makes them unique.

I also think that it will be fascinating to see all of my bits of DNA. This is the stuff that is a big part of making me who I am. (source: non-company website, Barry Starr, 23andMe: http://www.kqed.org/quest/blog/2009/07/20/taking-the-plunge-diving-into-my-dna/, accessed 24 September 2010)

Not all of these questions can be answered, but I am utterly curious about what makes me unique (or the eternal optimist, yet neurotic at times)? (source: company website, Jill Asher, 23andMe: https://www.23andme.com/pregnancy/thread/718/, accessed 24 September 2010)

So I do want to know as much about myself as I can. But more than that, I want to know about how I came to be who I am. (source: company website, Nancy Rabinowitz-Friedman, 23andMe: https://www.23andme.com/pregnancy/thread/708/, accessed 24 September 2010)

Theme 3: genealogy-related motivations and expectations

Various customers expressed an interest in tracing ancestors and were interested in knowing where they come from. Some held an interest in genealogy for a long time before they decided to purchase DTC GWT. These customers considered DTC GWT as a new technology that could enhance their understanding of genealogy and help in tracing their potential relatives.

Like many Americans, I don't have a clear picture of my geographically history… It will be fascinating—and a gift, really—to be able to say with a bit of clarity, "Well, according to my genome, I'm from…" (source: company website, Cecily Kellogg, 23andMe: https://www.23andme.com/pregnancy/thread/698/, accessed 24 September 2010)

They test autosomal markers for ancestry and health purposes. This means for your genealogy you can now match testers who are not on just your Ydna or your mtDNA lines. You can match people who are anywhere in your pedigree charts. (source: non-company website, Emily Aulicino, 23andMe: http://genealem-geneticgenealogy.blogspot.com/2009/12/23andme-success-story.html, accessed 24 September 2010)

I admit it. I have no self-discipline when it comes to genetic genealogy. When deCODEme launched, I had to be one of the first in line to get tested. (source: non-company website, Megan Smolenyak Smolenyak, deCODEme: http://www.rootstelevision.com/blogs/megans-rootsworld/2007/12/a_first_look_at_decodeme_dna_r.html, accessed 24 September 2010)

Theme 4: motivations and expectations related to participation in or contributing to research

Various users expressed an interest to donate their samples for future research.

"23andMe Sponsored Research: We will analyze your genetic and other voluntarily contributed personal information as part of our scientific research with the purpose of advancing the field of genetics and human health." I think many service customers will see this as a bonus. We want to volunteer, as it makes us feel we are part of something greater than ourselves. (source: non-company website, Grant Wood, 23andMe: http://exploringmygenes.blogspot.com/2009/06/reading-about-consents-risks-and.html, accessed 24 September 2010)

And the pregnancy community at 23andMe will give me an opportunity to contribute to future research on

> pregnancy conditions, discomforts and other health issues. (source: company website, Meagan Francis, 23andMe: https://www.23andme.com/pregnancy/thread/710/, accessed 24 September 2010)
> Or even that I may help someone else by contributing to research that could lead to breakthroughs in any number of medical fields. (source: company website, Nancy Rabinowitz-Friedman, 23andMe: https://www.23andme.com/pregnancy/thread/708/, accessed 24 September 2010)

Theme 5: recreation-related (non-genealogy-related) motivations and expectations

Various customers are interested in DTC GWT for recreational purposes other than formal genealogy. In particular, some users humorously expressed an interest in finding out if they could be related to someone famous. Of course, this was never stated as the main reason for purchasing a test, but it was mentioned as an "added bonus."

> Also, I'm hoping I'll find out that I'm distantly related to George Clooney - and that he'll want to meet me and then maybe we'll become friends, and then he'll fall madly in love with me and.... (source: company website, Nancy Rabinowitz-Friedman, 23andMe: https://www.23andme.com/pregnancy/profile/NYNancy/, accessed 24 September 2010)
> Impressed by Andrew Niccols' prescient (if insufficiently palindromic) GATTACA I assumed that there might even be SNPs that would convince Uma Thurman to have my babies. (source: non-company website, Myles Axton, deCODEme: http://blogs.nature.com/ng/freeassociation/2007/12/everything_you_need_to_last_yo.html, accessed 24 September 2010)

Discussion

The results of this exploratory study show that, overall, the major motivations and reasons for purchasing DTC GWT, as expressed by users, are linked to health. The fact that this theme is pervasive in the stories published on company websites whereas the stories from non-company websites express predominantly motivations related to curiosity and genealogy may suggest that companies are selectively publishing stories that promote health-related reasons. We cannot rule out the possibility that this is also partly due to the health focus of customers who decide to share their stories with companies. Nonetheless, the overall results are in line with other studies that have shown that health-related issues are at the center of individuals' reasons for wanting DTC genetic testing. This was reported in the studies by McGowan et al. (2010) and Kaufman et al. (2010), which, like our study, were focused on actual users or customers of DTC personal genome scanning services. Bloss et al. (2010) also showed that more than 80% of their study participants, who did purchase DTC GT but at a reduced price and within the context of a research project, wanted to know their genetic risk for treatable as well as for non-preventable disorders. Results from McGuire et al.'s (2009) survey, which addressed a more general audience of facebook-using individuals, suggest that of those who *have* used a personal genome test, 60% (38/60) consider the information obtained to be a diagnosis of medical condition or disease, and 51% used the test "To see if a specific disease runs in family or is in DNA." Finally, a recent study by Cherkas et al. (2010), in which a closed-ended survey was administered to adult twins participating in the TwinsUK Adult Twin Registry, showed that of those respondents who stated some interest in purchasing a "personal genetic screen," the most popular reason for doing so would be to "encourage me to adopt a healthier lifestyle if found to be at high risk of disease." Similarly, a large portion of respondents would obtain such testing so that their "doctor can monitor my health more closely."

The attention to health-related concerns from stories of users on company websites as well as those found on non-company websites is of particular interest as most DTC genetic testing companies have disclaimers on their websites stating that their services are not meant to be used as medical advice or as a diagnostic tool. For example, 23andMe states in the terms of service section of their website "23andMe Service Is For Research and Educational Use Only. We Do Not Provide Medical Advice, And The Services Cannot Be Used For Health Ascertainment or Disease Purposes" (23andMe 2010). Regarding deCODE's personal genome testing service (known as deCODEme), one can find in their service agreement and informed consent section "The Genetic Scan product is for informational purposes only, is not medical advice, and is not a substitute for professional medical advice, genetic counseling, diagnosis, or treatment" (deCODE 2010). The presence of these disclaimers is interesting for many reasons (Howard and Borry 2009), but we point out two principal issues. Firstly, it appears somewhat contradictory for companies to be posting these disclaimers and yet have users' stories on their websites focus mostly on the health-related aspects of the tests. Secondly, as all stories analyzed in our study come from people who have actually purchased tests, they should have, therefore, seen the companies' websites and signed the terms of services or informed consent forms. With this in mind, it is discon-

certing to see that some users may have overestimated the potential value of the tests purchased. Like us, McGowan et al. (2010) found that the two most common reasons for purchasing GWT were health related. However, they also revealed that half of their 23 interviewees "felt that the results of the scan are not medically actionable," and few participants changed their behavior after obtaining the results of the GWT. Furthermore, similarly to DTC GT companies, McGowan et al. (2010) distinguish the ideas of "informational value" from "medical value" stating that early adopters of GWT attribute a "largely informational rather than medical value" to this service. This differentiation as well as the distinction between medical information and health-related information may be meaningful in theory (especially legally); however, in practice, these demarcations may lose some of their significance.

In addition, it was noticed that some users did express an expectation of resolving their concerns regarding familial medical conditions based on the genetic test results. However, since we focused this study on motivations and expectations, we did not further analyze these expectations. (For example, were they realistic expectations? What was the apparent degree of (mis)understanding of the users regarding the information provided by the DTC genetic testing companies, including the test results?) Indeed, the study of consumer (mis)understanding of information remains an important aspect that requires further research.

Thus, overall, the information above tends to support the critiques of DTC genetic testing companies regarding the notion that consumers may be vulnerable to false or misleading advertising claims or to claims that tend to overstate the value of the companies' services in order to increase their sales (United States Government Accountability Office 2006; United States Government Accountability Office 2010). At the base of this problem is consumer/user understanding; how much do consumers and users understand about DTC genetic testing in general? Specifically, to what extent do recipients of GWT-based risk information understand their results, including the probabilistic nature of the risk estimates provided? Is there a risk that consumers may overestimate their value (McBride et al. 2010)? Some authors have argued that consumers may lack sufficient health literacy to interpret the genetic information provided (McBride et al. 2010). Moreover, Lachance et al. (2010) reported that the average reading grade level of DTC genetic testing websites is far above the average reading skill level of adults in the USA. That being said, McGowan and co-authors (2010) concluded that the early adopters they interviewed consider GWT "with both optimism and personal interest…and skepticism about the genomic technology's current capabilities." (McGowan et al. 2010) Being skeptical, however, does not imply having a great understanding of the technology or results thereof, and McGowan and co-authors do not report having studied users' outright understanding of GWT.

The potential impact of providing disease risk information generated from genomic test results on individual behavior and future health is also a matter of debate (Gulcher and Stefansson 2010; Ransohoff and Khoury 2010). There is presently little research regarding how individual consumers are likely to respond to their results. It is unclear whether this information could lead users to make positive health behavior changes, adopt more fatalistic attitudes, or increase/decrease anxiety levels (Bloss et al. 2010). A recent study by Bloss and colleagues (Bloss et al. 2011), however, does shed some light on this issue. They found no significant changes in anxiety levels, dietary fat intake, or exercise behavior after uptake of DTC GWT (from Navigenics Health Compass) in 2,037 research participants. The large majority of participants did not exhibit test-related distress, and there was no significant increase in the use of consequent follow-up screening tests following the test. Furthermore, little is known on how consumers might respond to changes in risk predictions and whether continual updates of risk predictions might change their perception of risk (Mihaescu et al. 2009). As stated earlier, however, McGowan et al's study did reveal that few early adopters used the information obtained from GWT to change their health-related behaviors.

The fact that our study focuses on the motivations and expectations of users means that, by default, we emphasize their interest in these services. However, it remains unclear to what extent the public at large is interested in these services and if the present model of DTC genetic testing is a sustainable one. In recent months, various DTC companies have ceased their activities (Borry et al. 2010a). One genome-wide-testing service we focused on for this study (which was in service when we started our study) was offered by Biomarker Pharmaceutical under the product name Gene Essence; the Gene Essence website with information regarding a GWT service, however, is no longer active. Other companies, like DNA Direct, Counsyl, and Navigenics, have changed their delivery model and now require consumers to order test via a medical doctor (Borry et al. 2010b). Foreshadowing these changes, a report by the investment bank Burril & Company (San Francisco) revealed that consumers' attachment to their physicians has been underestimated and that physicians remain the most likely source to which individuals will turn for health and genetic information (Burril & Company/Change Wave Research 2008). Furthermore, a recent calculation by Wright and Gregory-Jones (2010) estimates the current demand for DTC whole genome scans to be relatively small.

The willingness to participate in research, as expressed in the users' personal stories, mirrors the willingness of

many research volunteers. Numerous studies have indicated that many people are willing to permit the use of their tissue and information in a research project (Mezuk et al. 2008; Kettis-Lindblad et al. 2007). An important question with respect to a commercial setting, however, is whether customers are fully aware that they are participating in research. Based on a recent analysis of websites from five companies offering genome-wide testing directly to consumers, there is little evidence that the participation of customers in research is fully informed (Howard et al. 2010). As for the other three themes identified through users' personal stories: genealogy, curiosity/fascination, and recreation, these also overlap with reasons given by interviewees in McGowan's study (McGowan et al. 2010). (However, the authors did not discuss all these reasons in their present article, explaining that these reasons "fell outside the scope of" their analysis.) One motivation to purchase GWT identified through the latter's semi-structured interviews that was not identified as an important theme through personal users' stories is social networking possibilities. In other respects, it is interesting to note that a study of personal stories posted on non-company websites and company websites identified many of the same motivations, albeit without obtaining the same depth of information, as did McGowan's semi-structured interview study. That being said, we realize that it is possible that the interviewees in McGowan's study are also the authors of some of the stories we studied.

Foremost, our study provides concrete information regarding what motivates customers to purchase or use DTC genome-wide testing. However, there are some limitations with regard to methodology and the generalization of results that should be noted. Firstly, it must be acknowledged that since we accessed users based on the posting of their stories on blogs or company websites, their views may not be representative of all other users who purchased DTC GWT (but who did not post their stories). Secondly, as many personal stories emanate from companies' websites, it is not clear whether these narratives are genuine stories of users or if they were constructed in collaboration with companies in order to publish archetype stories as part of public relations' initiative. Moreover, even if these are genuine users' stories, it is expected that companies would have screened the stories in some way. Thirdly, as our research was focused on companies offering genome-wide testing, the results are not necessarily applicable to other types of DTC genetic testing companies, which are selling, for example, single-gene or multiple-gene tests. This is an important point because many DTC genetic testing companies do not sell GWT, and issues raised and conclusions made about the latter do not always apply to the former. As far as the scope of this study is concerned, it was focused on personal stories of customers who underwent testing; it does not report information regarding consumers who have not purchased tests. Moreover, concerns or misgivings regarding DTC genetic tests or genetic testing in general were not studied.

Finally, an important question for users remains unanswered: "why purchase a genetic test from a company instead of obtaining it through the traditional health care (THC) system?" Unfortunately, the answer to this question would not be informative with regard to genome-wide testing as this service is not (yet) offered via the THC system. As for companies offering single-gene or multiple-gene testing, they have largely been neglected in (academic) studies and debates. Therefore, all studies up until now which have focused on DTC personal genome services are really asking "why do you want to have hundreds of thousands or millions of genetic markers tested?" The "direct-to-consumer" aspect is somewhat secondary since there is no real alternative source to obtain these services. This has caused some confusion because it is not the same as asking users why they opted for the commercial offer of a test they could have obtained via the THC system (i.e., BRCA1 testing). This question, however, is important if the THC system aims to offer quality (and by default, useful) genetic testing for the public. Here, the notion of genetic testing as a consumer product or a health care service becomes paramount.

Conclusions

Little is known about the motivational factors that stimulate individuals to use DTC genome-wide testing. This article is only the second study focused on studying actual users' perspectives; we reported five major sets of motivations relating to health, curiosity/fascination, genealogy, contributing to research, and recreation. The exploratory character and results of this study show the need for further research in order to better understand users' motivations to use DTC genome-wide testing and DTC genetics tests in general. Further studies should include the understanding of users with respect to the material on company websites as well as the character and nature of this information (i.e., is it based on scientific references, misleading, promotional, confusing?). Further attention should also be paid to how users' motivation may change with time, how it may be related to specific disease experiences, and other educational/professional, familial, interpersonal, and social factors. Such information may help us understand how users help shape the commercial market of DTC GT and more importantly, if such testing is truly helpful in improving their health status. Although it can be a difficult task to obtain meaningful public feedback, it is a crucial task. Knowing what the public wants is important, especially for commercial endeavors, but knowing what the public needs to improve their health can be a completely different question. If

offering genomic information to improve health is truly our goal, then we must make certain not confuse the two issues.

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References

23andMe (2010) Terms of service. https://www.23andme.com/about/tos/ Accessed 11 Nov 2010

Allison M (2010) Genetic testing clamp down. Nat Biotechnol 28:633

American College of Obstetricians and Gynaecologists (2008) ACOG Committee Opinion no. 409: direct-to-consumer marketing of genetic testing. Obstet Gynecol 111:1493–1494

Bloss CS, Ornowski L, Silver E, Cargill M, Vanier V, Schork NJ, Topol EJ (2010) Consumer perceptions of direct-to-consumer personalized genomic risk assessments. Genet Med 12:556–566

Bloss CS, Schork NJ, Topol EJ (2011) Effect of direct-to-consumer genomewide profiling to assess disease risk. New Engl J Med 364 (6): 524–534

Borry P, Howard HC, Senecal K, Avard D (2009a) Direct-to-consumer genome scanning services. Also for children? Nat Rev Genet 10:8

Borry P, Howard HC, Senecal K, Avard D (2009b) Health-related direct-to-consumer genetic testing: a review of companies' policies with regard to genetic testing in minors. Fam Cancer 9:51–59

Borry P, Cornel MC, Howard HC (2010a) Where are you going, where have you been. Direct-to-consumer genetic tests for health purposes. J Community Genet 1:101–106

Borry P, Henneman L, Lakeman P, ten Kate LP, Cornel MC, Howard HC (2010b) Preconceptional carrier genetic testing and the commercial offer directly-to-consumers. Hum Reprod 26:972–977

Cherkas LF, Harris JM, Levinson E, Spector TD, Prainsack B (2010) A survey of UK public interest in internet-based personal genome testing. PLoS ONE 5:e13473

Collins F (2010) The language of life: DNA and the revolution in personalized medicine. Harper Collins, New York

Burril & Company/Change Wave Research (2008) Personalized medicine and wellness survey. Executive summary. http://www.burrillandco.com/content/CWSurvey_61708.pdf. Accessed 21 Sept 2010

deCODE (2010) deCODEme Genetic scan service agreement and informed consent. http://www.decodeme.com/service-agreement Accessed 11 Nov 2010

Duncan D (2009) The experimental man. Wiley, Hoboken

European Society of Human Genetics (2010) Statement of the ESHG on direct-to-consumer genetic testing for health-related purposes. Eur J Hum Genet 18:1271–1273

Federal Trade Commission (2009) FTC facts for consumers. At-home genetic tests: a healthy dose of skepticism may be the best prescription. http://www.ftc.gov/bcp/edu/pubs/consumer/health/hea02.shtm. Accessed 26 Apr 2011

Foster MW, Royal CDM, Sharp RR (2006) The routinisation of genomics and genetics: implications for ethical practices. J Med Ethics 32:635–638

Foster MW, Mulvihill JJ, Sharp RR (2009) Evaluating the utility of personal genomic information. Genet Med 11:570–574

Gulcher J, Stefansson K (2010) Genetic risk information for common diseases may indeed be already useful for prevention and early detection. Eur J Clin Investig 40:56–63

Hogarth S, Javitt G, Melzer D (2008) The current landscape for direct-to-consumer genetic testing: legal, ethical, and policy issues. Annu Rev Genomics Hum Genet 9:161–182

Howard HC, Borry P (2009) Personal genome testing: do you know what you are buying? Am J Bioeth 9:11–13

Howard HC, Knoppers BM, Borry P (2010) Blurring lines. The research activities of direct-to-consumer genetic testing companies raise questions about consumers as research subjects. EMBO Rep 11:579–582

Human Genetics Commission (2010) A common framework of principles for direct-to-consumer genetic testing services. http://www.hgc.gov.uk/Client/document.asp?DocId=280&CAtegoryId=10 Accessed 17 Feb 2011

Hunter DJ, Khoury MJ, Drazen JM (2008) Letting the genome out of the bottle—will we get our wish? N Engl J Med 358:105–107

Janssens AC, Gwinn M, Bradley LA, Oostra BA, van Duijn CM, Khoury MJ (2008) A critical appraisal of the scientific basis of commercial genomic profiles used to assess health risks and personalize health interventions. Am J Hum Genet 82:593–599

Kaufman D, Murphy Bollinger J, Devaney S, Scott J (2010) Direct from consumers: a survey of 1,048 customers of three direct-to-consumer personal genomic testing companies about motivations, attitudes, and responses to testing.http://www.ashg.org/cgi-bin/2010/showdetail.pl?absno=21043 Accessed 26 Apr 2011

Kettis-Lindblad A, Ring L, Viberth E, Hansson MG (2007) Perceptions of potential donors in the Swedish public towards information and consent procedures in relation to use of human tissue samples in biobanks: a population-based study. Scand J Public Health 35:148–156

Khoury MJ (2003) Genetics and genomics in practice: the continuum from genetic disease to genetic information in health and disease. Genet Med 5:261–268

Lachance CR, Erby LA, Ford BM, Allen VC Jr, Kaphingst KA (2010) Informational content, literacy demands, and usability of websites offering health-related genetic tests directly to consumers. Genet Med 12:304–312

McBride CM, Alford SH, Reid RJ, Larson EB, Baxevanis AD, Brody LC (2009) Characteristics of users of online personalized genomic risk assessments: implications for physician–patient interactions. Genet Med 11:582–587

McBride CM, Wade CH, Kaphingst KA (2010) Consumers' views of direct-to-consumer genetic information. Annu Rev Genomics Hum Genet 11:427–446

McGowan ML, Fishman JR, Lambrix MA (2010) Personal genomics and individual identities: motivations and moral imperatives of early users. New Genet Soc 29:261–290

McGuire A, Diaz CM, Wang T, Hilsenbeck S (2009) Social networkers' attitudes toward direct-to-consumer personal genome testing. Am J Bioeth 9:3–10

Mezuk B, Eaton WW, Zandi P (2008) Participant characteristics that influence consent for genetic research in a population-based survey: the Baltimore epidemiologic catchment area follow-up. Community Genet 11:171–178

Mihaescu R, van Hoek M, Sijbrands EJ, Uitterlinden AG, Witteman JC, Hofman A, van Duijn CM, Janssens AC (2009) Evaluation of risk prediction updates from commercial genome-wide scans. Genet Med 11:588

Nuffield Council on Bioethics (2010) Medical profiling and online medicine: the ethics of 'personalised healthcare' in a consumer age. Nuffield Press, Oxfordshire

Ransohoff DF, Khoury MJ (2010) Personal genomics: information can be harmful. Eur J Clin Invest 40:64–68

Richards M (2010) Reading the runes of my genome: a personal exploration of retail genetics. New Genet Soc 29:291–310

Silverman D (2006) Interpreting qualitative data. Sage, London

United States Government Accountability Office (2006) Nutrigenetic testing: tests purchased from four websites mislead consumers. US GAO, Washington

United States Government Accountability Office (2010) Direct-to-consumer genetic tests misleading test results are further complicated by deceptive marketing and other questionable practices .http://energycommerce.house.gov/documents/20100722/Kutz.Testimony.07.22.2010.pdf. Accessed 10 Aug 2010

Wade CH, Wilfond BS (2006) Ethical and clinical practice considerations for genetic counselors related to direct-to-consumer marketing of genetic tests. Am J Med Genet C Semin Med Genet 142:284–292

Wilde A, Meiser B, Mitchell PB, Schofield PR (2010) Public interest in predictive genetic testing, including direct-to-consumer testing, for susceptibility to major depression: preliminary findings. Eur J Hum Genet 18:47–51

Wright CF, Gregory-Jones S (2010) Size of the direct-to-consumer genomic testing market. Genet Med 12:594